



ARID1B gene

AT-rich interaction domain 1B

Normal Function

The *ARID1B* gene provides instructions for making a protein that forms one piece (subunit) of several different SWI/SNF protein complexes. SWI/SNF complexes regulate gene activity (expression) by a process known as chromatin remodeling. Chromatin is the network of DNA and protein that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling is one way gene expression is regulated during development; when DNA is tightly packed, gene expression is lower than when DNA is loosely packed.

Through their ability to regulate gene activity, SWI/SNF complexes are involved in many processes, including repairing damaged DNA; copying (replicating) DNA; and controlling the growth, division, and maturation (differentiation) of cells. The ARID1B protein and other SWI/SNF subunits are thought to act as tumor suppressors, which keep cells from growing and dividing too rapidly or in an uncontrolled way.

The ARID1B subunit is able to attach (bind) to DNA and is thought to help target SWI/SNF complexes to the chromatin location that needs to be remodeled.

Health Conditions Related to Genetic Changes

Coffin-Siris syndrome

At least nine mutations in the *ARID1B* gene have been found to cause Coffin-Siris syndrome. This condition is characterized by delayed development, abnormalities of the fifth (pinky) fingers or toes, and characteristic facial features that are described as coarse. Most *ARID1B* gene mutations involved in Coffin-Siris syndrome lead to an abnormally short, nonfunctional protein. As a result, affected individuals have half the normal amount of functional ARID1B protein. Although it is unclear how these changes affect SWI/SNF complexes, researchers suggest that *ARID1B* gene mutations result in abnormal chromatin remodeling. Disturbance of this process alters the activity of many genes and disrupts several cellular processes, which could explain the diverse signs and symptoms of Coffin-Siris syndrome. People with Coffin-Siris syndrome do not appear to have an increased risk of cancer (see below).

cancers

Mutations in the *ARID1B* gene are involved in several types of cancer, including breast cancer, a childhood cancer of nerve tissue called neuroblastoma, and a type

of blood cancer called diffuse large B-cell lymphoma. These mutations are somatic, which means they are acquired during a person's lifetime and are present only in tumor cells. The mechanism by which mutations in the *ARID1B* gene contribute to cancer is unknown, although it is thought that changes in SWI/SNF complexes are involved. These changes may impair normal cell differentiation, which leads to the overgrowth of certain cell types, causing cancer. Alternatively, abnormal SWI/SNF complexes may disrupt the regulation of genes that help control the growth and division of cells, which leads to cancer. It is likely that other genetic changes in addition to *ARID1B* gene mutations are necessary for cancer development.

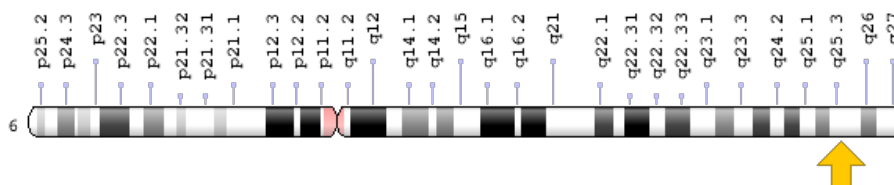
other disorders

Mutations in the *ARID1B* gene can cause intellectual disability without other hallmark features of Coffin-Siris syndrome. As in Coffin-Siris syndrome, the gene mutations that cause intellectual disability lead to an abnormally short, nonfunctional protein, and affected individuals have half the normal amount of ARID1B protein. It is unclear why some people with an *ARID1B* gene mutation develop intellectual disability and others have the additional features of Coffin-Siris syndrome.

Chromosomal Location

Cytogenetic Location: 6q25.3, which is the long (q) arm of chromosome 6 at position 25.3

Molecular Location: base pairs 156,777,847 to 157,210,779 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 6A3-5
- ARI1B_HUMAN
- ARID domain-containing protein 1B
- AT rich interactive domain 1B (SWI1-like)
- AT-rich interactive domain-containing protein 1B

- BAF250B
- BRG1-associated factor 250b
- BRG1-binding protein ELD/OSA1
- BRIGHT
- DAN15
- ELD (eyelid)/OSA protein
- ELD/OSA1
- KIAA1235
- MRD12
- OSA2
- P250R

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): ATP-Driven Chromatin Remodeling Machines Change Nucleosome Structure
<https://www.ncbi.nlm.nih.gov/books/NBK26834/#A644>
- Molecular Biology of the Cell (fourth edition, 2002): Chromosomal DNA and Its Packaging in the Chromatin Fiber
<https://www.ncbi.nlm.nih.gov/books/NBK26834/>
- Simons VIP Connect: Single Gene - ARID1B
https://www.simonsvipconnect.org/index.php?option=com_content&view=article&id=541&catid=90&Itemid=492&lang=en

GeneReviews

- Coffin-Siris Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK131811>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ARID1B%5BTIAB%5D%29+OR+%28AT+rich+interactive+domain+1B%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- AT-RICH INTERACTION DOMAIN-CONTAINING PROTEIN 1B
<http://omim.org/entry/614556>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ARID1B.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ARID1B%5Bgene%5D>
- HGNC Gene Family: AT-rich interaction domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/418>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=18040
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/57492>
- UniProt
<http://www.uniprot.org/uniprot/Q8NFD5>

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